Inborn Errors of Metabolism

GLYCOGEN STORAGE DISEASES

MUSCLE GLYCOGENOSES

Muscle glycogen phosphorylase ***(2.4.1.1)***
GSD Type V McArdle Disease
MIM 232600

LIVER GLYCOGENOSES

Amylo-1,6-glucosidase ***(3.2.1.33)***
Glucanotransferase ***(2.4.1.25)***
GSD Type III Cori Disease
MIM 232400

Glucose-6-phosphatase ***(3.1.3.9)***
GSD Type I Von Gierke Disease
MIM 232200

MONO- & DI-SACCHARIDES

Sucrase-isomaltase ***(3.2.1.10)***
Sucrose-Isomaltose Malabsorption
MIM 609845

Lactase ***(3.2.1.23) (3.2.1.108)***
Lactose Intolerance
MIM 223000

Galactokinase ***(2.7.1.6)***
Galactose Kinase Deficiency
MIM 230200

Galactose-1-phosphate uridyl transferase ***(2.7.7.12)***
Classical Galactosaemia
MIM 606999

PEROXISOMAL DEFECTS

Alanine-glyoxylate aminotransferase ***(2.6.1.44)***
Hyperoxaluria Type 1
MIM 259900

LIPID METABOLISM

Hydroxymethylglutaryl CoA lyase ***(4.1.3.4)***
Ketone Synthesis Defect
MIM 246450

Medium chain acyl CoA dehydrogenase ***(1.3.99.3)***
MCAD Deficiency
MIM 607008

SPHINGOLIPIDOSES

Acid-β-glucosidase ***(3.2.1.45)***
Gaucher Disease
MIM 230800

Hexosaminidase A ***(3.2.1.52)***
Tay Sachs Disease
MIM 272800

Sphingomyelinase ***(3.1.4.12)***
Niemann-Pick Disease
MIM 257200

RESPIRATORY CHAIN DEFECTS

Cytochrome c oxidase ***(1.9.3.1)***
Complex IV
MIM 220110

PORPHYRIAS

Porphobilinogen deaminase ***(2.5.1.61)***
Acute Intermittent Porphyria
MIM 176000

Uroporphyrinogen decarboxylase ***(4.1.1.37)***
Porphyria Cutanea Tarda
MIM 176100

Protoporphyrinogen oxidase ***(1.3.3.4)***
Variegate Porphyria
MIM 176200

AMINO ACID METABOLISM

Fumarylacetoacetase ***(3.7.1.2)***
Tyrosinaemia Type I
MIM 276700

Phenylalanine hydroxylase ***(1.14.16.1)***
Phenylketonuria (PKU)
MIM 261600

Tyrosinase ***(1.14.18.1)***
Tyrosine Negative Albinism
MIM 203100

Glycine dehydrogenase ***(1.4.4.2)***
Non Ketotic Hyperglycinaemia (NKH)
MIM 238300

PURINE & PYRIMIDINE METABOLISM

Xanthine oxidase ***(1.17.3.2)***
Xanthinuria
MIM 278300

Hypoxanthine guanine phospho-ribosyl transferase ***(2.4.2.8)***
HGPRT Deficiency, Primary Gout, Lesch-Nyhan Syndrome
MIM 308000

Adenosine deaminase ***(3.5.4.4)***
Severe Combined Immuno-Deficiency (SCID)
MIM 102700

FOLIC ACID

Dihydrofolate reductase ***(1.5.1.3)***
Megaloblastic Anaemia
MIM 126060

ORGANIC ACIDURIAS

Propionyl-CoA carboxylase ***(6.4.1.3)***
Propionic Acidaemia
MIM 606054

Methylmalonyl-CoA mutase ***(5.4.99.2)***
Methylmalonic Acidaemia
MIM 251000

Isovaleryl-CoA dehydrogenase ***(1.3.99.10)***
Isovaleric Acidaemia
MIM 243500

AMINO ACID METABOLISM

Cystathionine synthetase ***(4.2.1.22)***
Homocystinuria
MIM 236200

Branched chain ketoacid decarboxylase ***(1.2.4.4)***
Maple Syrup Urine Disease
MIM 248600

GABA METABOLISM NEUROTRANSMITTER DEFECTS

Succinic semialdehyde dehydrogenase ***(1.2.1.24)***
4-Hydroxybutyric Aciduria
MIM 271980

4-Aminobutyrate aminotransferase ***(2.6.1.19)***
GABA Transferase Deficiency
MIM 137150

UREA CYCLE DEFECTS (HYPERAMMONAEMIA)

Carbamoyl phosphate synthase ***(6.3.4.16)***
CPS Deficiency
MIM 237300

Ornithine carbamoyl transferase ***(2.1.3.3)***
OCT Deficiency
MIM 311250

Argininosuccinate synthase ***(6.3.4.5)***
Citrullinaemia
MIM 215700

Argininosuccinate lyase ***(4.3.2.1)***
Argininosuccinate Aciduria
MIM 207900

Designed by Donald Nicholson, The University of Leeds, England – and Sigma-Aldrich
in collaboration with Mick Henderson, St.James' Hospital, Leeds – and the UK MetBioNet Training Group *www.metbio.net.*

For further information about metabolic disorders visit the OMIM (Online Mendelian Inheritance in Man) web site, hosted by the John Hopkins University – *www.ncbi.nlm.gov/entrez/query/fcgi?db=OMIM*

GDS